



Noah and Laine VanHoutan: An Unbreakable Bond

A Special Family: The VanHoutans

Reprinted with permission from Taylor's Tale December 2011 Newsletter at www.taylorstale.com.

Noah VanHoutan was like any other 3-year-old. Fearless and full of energy, Noah didn't think twice about taking risks like jumping off the playground slide. He loved bats, balls and trains and being a protective big brother for his twin sisters, Laine and Emily.

But when Noah began experiencing delayed speech and small tremors, his parents grew worried. They took him to a doctor who reassured them and said Noah's behavior and development were typical. Eight months later, Noah stopped breathing and collapsed in a scary episode doctors later determined to be a seizure, but EEG and MRI results were normal. Nevertheless, he was diagnosed with childhood epilepsy. Doctors were puzzled again when, almost a year later, an MRI scan showed signs of brain atrophy. Noah's

speech was regressing, and he had difficulty performing simple tasks like brushing his teeth or eating with a fork. It would take nearly two years and several neurologists before Noah was diagnosed with a rare and fatal genetic disorder—late infantile NCL, a form of Batten disease.

Tracy and Jennifer VanHoutan were devastated and still coping with their son's diagnosis when they learned exactly five months later that their daughter Laine also inherited the incurable disease. Though symptom-free at the time, within weeks, the little girl who adored tutus and playing dress-up suffered her first seizure. While Laine's speech, mobility and vision began to decline, her twin sister Emily—the only sibling to avoid inheriting the disease—made new

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A SPECIAL FAMILY (CONTINUED FROM PAGE 1)

friends and learned new skills. An ever-present reminder of the life Laine could have had, the gap between the two continues to grow with time.

At 5 years old, today Laine is the same determined child as when she was a baby. Although her vocabulary is shrinking, she uses body language to communicate. Instead of reading stories out loud, she now quietly flips through her books. But she still smiles all the time and blows kisses to her mom and dad from across the room. For Noah, who is now 7, the disease has progressed even more. Confined to a wheelchair, Noah can't walk or talk and uses a feeding tube. As brother and sister joined by their shared struggle, Noah and Laine share a special bond. Noah is the first person Laine wants to see when she wakes up and when she gets off the school bus. The two spend a great deal of time together, and Laine often comforts her brother by holding his hand, especially when he's having a meal or being given medicine.

The VanHoutans cherish the time they have and frequently go on vacation together, including trips to Disney World, Washington, DC and Colorado. A family skiing trip in 2010 is a particularly cherished memory for them. Noah cruised down the mountain in a sled, while Emily and Laine skied. At that time, Laine was still able to go up the slope all by herself in her boots and skis. Just a year later, the progression of the disease has robbed her of such abilities.

After Noah's diagnosis, Tracy and Jennifer began Noah's Hope as a way to raise awareness and funds to find a cure for their son. Little did they know that they would soon seek the same solution for their daughter. What began as a grassroots effort has blossomed into a larger initiative that's raised more than \$250,000 to support researchers and the Batten Disease Support and Research Association (BDSRA). Presently, educating lawmakers and voters on proposed legislation that will impact rare disease research is a major focus for Noah's Hope, and they hope to involve other

Batten disease non-profits in their activism efforts.

As a word of advice for families fighting Batten and other life-threatening diseases, Jennifer and Tracy say it's important to savor every moment. "Be your own advocate and live without regrets," Tracy added.

To learn more, visit noahshope.com.

Annual Membership Dues

Annual membership dues are payable beginning in January 2012. The BDSRA national headquarters is your most vital link to information, support, and encouragement for families with children who have Batten disease. BDSRA is also the largest private source of funds for international research on Batten disease.

We ask each individual to pay \$40 for annual dues at the beginning of each new year. We hope you will continue your membership with BDSRA, or perhaps join for the first time.

Please make payment directly to BDSRA and not the Chapter affiliations. If you have any questions, please contact us at bdsra1@bdsra.org. Your support and participation is greatly appreciated!



Pride of Ireland!

Contributed by Tony Heffernan

Three year-old Liam Heffernan, who is affected by Batten disease, became Ireland's youngest recipient of 2011's National Child of Courage Award. Liam, of Castledrum, Keel Castlemaine, County Kerry, attended the Gala Black Tie event in Limerick on November 26 accompanied by parents Tony and Mary and his usual small collection of dinosaurs. Liam accepted his award to a standing ovation of more than 300 supporters of the Share A Dream Ireland annual event.

Following a special mention by Miriam O'Callaghan of RTE, MC for the night, Will Leahy from RTE's 2FM, told Liam's story of the past twelve months, about his battle with Batten disease, and how he became the world's youngest child, at just 2 years old, to undergo a medical treatment trial in New York on May 3, 2011 to save his life. Leahy also told the crowd about the special relationship Liam had with his only sibling, sister Saoirse, who sadly passed away at the age of five in January 2011 from the same condition.

The Heffernan Family are awaiting news on Liam's results following a recent six month check-up at the New York hospital that carried out the eight-hour operation in May. They hope that no deterioration in Liam's brain mass has been observed, which will give hope that Liam's life may be extended.

In total, eight children received the award, and all eight children and their families were given trips to Euro Disney as part of their awards for being Ireland's Children of Courage.



National Child of Courage Award winner, Liam Heffernan, attends the Black Tie Gala in Limerick with (from left) Shay Kinsella, of Share a Dream Ireland, and Liam's parents, Mary and Tony Heffernan.

Little Brandon's Legacy Lives on in Charity Drive

Inspired by boy's battle with Batten disease

By Geoff Kirbyson, Winnipeg Free Press

A little boy who captured the hearts of Winnipeggers during his battle with Batten's disease has always been a hero to his family, but now he plays that same role for many other children facing health challenges of their own.

The proceeds from sales of a special necklace inspired by Brandon Smith will go toward a newborn screening program for cystic fibrosis at the Children's Hospital.



Jewelry designer Hilary Druxman created the "hero" necklaces to honour the memory of Brandon Smith, who died two years ago at age eight. (Photo by Ruth Bonneville/Winnipeg Free Press)

Designed by Hilary Druxman, the piece - a sterling silver disc pendant engraved with the word 'hero' and a small heart - was sold in support of the fifth-annual Ice Crystal Gala, a sold-out fundraiser held November 21 at the Winnipeg Convention Centre.

Brandon was diagnosed with Batten's disease, a fatal affliction that shrinks the brain, as an 18-month-old in late 2002. The life expectancy for those

with the disease is about five years but Brandon blazed his own trail and, through groundbreaking therapy and treatment, not to mention dogged determination, regularly astounded medical professionals for several years until he could fight no more. He died two years ago this month at the age of 8½.

"The fact that Brandon isn't being forgotten means the world to me," said his mom, Cindy Smith. "And that he's still able to give back even though he isn't still physically here is great for me as well."

Druxman said she was approached by the Children's Hospital Foundation about creating a piece of jewelry back in the spring. While waiting for inspiration to strike, Doug Smith walked in her door and asked her about making something for his family to remember Brandon by.

When she asked him what word should be put on the charm, he replied, "Hero, he was our hero."

After finishing the design, with a little help from Brandon's big sister, Hayley, Druxman called Smith to tell him she had also included a little heart on the disc. She didn't know at the time, but when the Smiths sign Christmas or birthday cards, they include a small heart after their three names in honour of Brandon.

The necklace sells for \$35, including tax, with 100 per cent of the proceeds going to the hospital's foundation. It's available online at www.hilarydruxman.com.

Lesia Sianchuk, executive director of the foundation, said it continues to sell the necklace now that the gala is over. "The spirit of Brandon living on through this piece is what we're embracing. The word 'hero' is a significant word for our foundation," she said.



The pendant features the word "hero" and a heart. (Photo by Ruth Bonneville/Winnipeg Free Press)

From the Executive Director

By Lance Johnston

As most of you may know, I have made the decision to step down as BDSRA's Executive Director soon after the first of the year. After leading BDSRA in some capacity since 1988, I have come to recognize that the organization needs someone with more skills and energy than I have.

BDSRA is poised to move forward into a new realm of support and research that will be beneficial to all. Having had no formal training to be a director, I have done the best I can,

but am smart enough to know when the time for change has come.

I will continue to be with BDSRA full time but in a different capacity, primarily working with families and their children and, hopefully, keeping in touch with research until the day comes when someone who is a scientist can take the lead in that area. It has been quite a journey, and one that I am proud to have been a part of, and will continue to be a part of -- the movement that will one day have a cure for this terrible disease.

The BDSRA Board of Directors has started the search for a new Executive Director and I will be here to help him/her get started and take the reins. I am excited at the prospect of having a new leader that will be able to keep BDSRA advancing.

All of us have had a hand in getting BDSRA to where we are today and we will all continue to keep the momentum going. Watch the website, *The Illuminator* and your mail for updates as they occur.

AND THEY'RE OFF... 2012 Annual Conference

Plans for BDSRA's Annual Conference are moving along! Please mark your calendars and plan to join us in Charlotte, North Carolina, July 19–22, 2012, at the Charlotte Marriott Executive Park. Conference registration materials and the program agenda will be in the April 2012 issue of *The Illuminator*.



The reservation link at the Marriott is now available. To make your reservations, you can either call (800) 359-7961 and refer to Batten Disease Support (or 3 letter group code: BTN) or cut/paste one of the links below to your web browser and enter your arrival/departure dates. The group rate of \$94.00 will already be entered.

<http://www.marriott.com/hotels/travel/CLTNC?groupCode=BTNBTNA&app=resvlink&fromDate=7/17/12&toDate=7/23/12>

<http://www.marriott.com/hotels/travel/cltnc-charlotte-marriott-executive-park/?toDate=7/23/12&groupCode=BTNBTNA&fromDate=7/17/12&app=resvlink>

If you have any questions or concerns, please address them to either Charlie Leffler, Conference Chair, at dresden1@mindspring.com, Lance Johnston at bdra1@bdra.org, or Nancy Carney at nancycarney@bdra.org.

From the Desk of the Director of Development

By Adina Ryan

Happy New Year!

I hope that everyone had a wonderful holiday season and was able to enjoy treasured time with family and friends. We have certainly enjoyed receiving all the holiday cards and photos of your families, visiting with some of the children and sharing in the joy of Christmas. Now we turn our thoughts to a brand new year with hopes for exciting progress in Batten disease research and in our

support of all of you with our programs and services.

As you receive this newsletter, I will no longer be the development director at BDSRA. I have received an opportunity to work closer to home where I can share more of my time with my family and my community. I am so grateful to have had the chance to meet so many of you and to play a small part in sharing your lives. Thank you for all your kindness to me these

last two years. You will all be forever embedded in my heart. Your cause is still my cause, and I hope that the next generation of leaders for BDSRA will be able to do even more to help eradicate this disease. I am just a Facebook page away and certainly hope that I can stay a part of your family. Best wishes to all of you for a very wonderful 2012.

With utmost respect and admiration,
Adina Ryan

Annual Fund Drive Sets Goal for \$125,000 to Support Programs and Services

Can you imagine

a day when this disease no longer exists? That is the goal for all of us. Unfortunately, in the meantime, we need to continue important research and programs that help our families learn more about the disease, educate teachers, network with other families, and provide services for fundraising, medical needs and testing. These are all areas where your donation to the BDSRA annual fund can provide support. Without your generosity, we cannot help you or others. Please consider making a donation in 2012 to our annual fund. You may make recurring monthly gifts by contacting us directly at (800) 448-4570 or by making a one-time donation online at www.bdsra.org.



Upcoming Events

Feb 29



June 1 - 3 (USA)



July 19 - 22



Fundraising News

Thanks to everyone who held a fundraiser this past quarter!

Together, over \$57,000 was raised to further the hope for a cure. Thanks for your support!



Celia's Walk



Averee's Purpose



Barkin' for Batten

Fundraising MATTERS!

Check out this quarterly idea corner to help in your efforts to raise funding for research, programs and support. This quarter's idea:

No-Show Events

Are you looking for a great fundraising idea, but have already been to one too many parties over the holidays, or don't want to trek through bad winter weather to organize an event? A no-show event is a perfect way to raise money with little effort. It's very simple - just invite people to a function and ask them NOT to show up!

No-show events like potlucks, golf outings, Super Bowl parties or bingo tournaments encourage people to donate instead of showing up to the event. It's a fun, creative and easy way to host a great fundraiser. Please contact us at (800) 448-4570 for more information about this type of event.

Don't forget some of our great **COMMUNITY PROGRAMS** at BDSRA to raise money every day without costing you a cent:

Cartridges for Kids — our recycling program for used electronics.

GoodSearch, Goodshop and NEW GoodDining — simply visit www.goodsearch.com and select Batten Disease Support and Research Association as your charity of choice, then search, shop or dine out in over 10,000 participating restaurants!

Capital One Connect — apply for our branded credit card and 1% of all your purchases made on the card are donated to us.



United Way and Combined Federal Campaign

Your United Way or Combined Federal Campaign contributions can benefit the work of BDSRA. Every fall, kick off campaigns occur in the workplace. Why not make your contributions benefit those in your life affected by Batten disease? Whether you are a postal worker, in the military, or just happen to work where United Way contributions are encouraged, you can designate our organization and help those who you love fighting for a cure with this disease. For further details, contact BDSRA.

BDSRA Programs — It's All About You!

By Lisa Weston, Program Director

Quarterly Reminders...

Our **Sibling Carrier Testing Program** is available to siblings of individuals affected with Batten disease to be tested for carrier status. The program will also cover carrier testing for qualified spouses or fiancées, and diagnostic testing for children of siblings. For more information regarding testing protocol, eligibility, costs and available funding, confidentiality, etc., please contact me.



The BDSRA **Equipment Exchange Program** is open to any BDSRA family needing equipment for their child/ren living with Batten disease. We have a variety of medical equipment and supplies, most of which have been generously donated by other families, and is available at no costs to the family. Please contact me if you are interested in the program, want to donate or have any questions.

Special Needs Planning

The following articles are reprinted with permission from *The Academy of Special Needs Planners' October 2011 Newsletter Special Needs Planning News*.

STARTING A NEW JOB? CONSIDER THESE TIPS WHEN SIGNING UP FOR EMPLOYEE BENEFITS

When you start a new job you may be offered a wide range of employee benefits, from retirement accounts to life insurance. If you are the parent of a child with special needs, the decisions you make when signing up for these benefits could have dramatic consequences well down the road. Here are some things to think about when it comes time to make those important employee benefit elections on your first day. (Of course, if you made different choices, there is always time to change your plan.)

Don't name a child with special needs as a direct beneficiary of a retirement account

When you first set up an IRA or 401(k), you have to name primary

and contingent beneficiaries to receive the funds in the account if you pass away. In most cases, your first thought will be to name your spouse or partner as the primary beneficiary and your children as the contingent beneficiaries. However, if one of your children has special needs, this decision could be disastrous for several reasons. First, if your child is receiving government benefits, a sudden influx of income from an inherited retirement account could ruin his access to those benefits. Second, most retirement accounts pay out a set amount of money per year to the designated beneficiaries, and this amount could be much more than a person with special needs can or should handle on his own. Finally, you may be able to set up a special needs trust to appropriately hold the retirement funds.

Evaluate how much life insurance you may need and designate the appropriate beneficiary

Many companies offer basic life insurance coverage as part of a benefits package, with the option to purchase additional insurance at a discount. As discussed above, naming a child with special needs as the beneficiary of a large life insurance policy could create havoc for the child later on. However, life insurance does provide a great planning opportunity for parents of children who may require significant care because the death benefit can be used to fund a special needs trust with a significant amount of money. If your company offers additional insurance, it may be prudent to purchase it and name a special needs trust as the beneficiary. Furthermore, if you are concerned about being "fair" to your children without special needs when it comes

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to inheritance, you may decide to leave them your retirement accounts while using a life insurance policy to take care of your child with special needs.

Long-term disability insurance may be worth it

Because families of children with special needs often face significant costs, you have to give serious consideration to what your family's financial position would be if you were to suddenly lose your job due to disability. Many companies allow employees to purchase long-term disability insurance and some offer it for free. If you are the primary wage earner and you don't have substantial savings to care for your family if you are disabled, then it may make sense to sign up for disability insurance from the get-go so that you know your family will be protected if something happens to you.

If you are starting a new job, or if it is time to review your employee benefits package at your current job, take a moment to go over your benefits with your special needs planner. You may be able to take advantage of benefits you never even knew you had.



SPECIAL NEEDS PLANNING IS A MARATHON, NOT A SPRINT

Unlike some areas of law like “employment discrimination law” or “patent law,” special needs planning does not focus on one specific legal principle or topic. Instead, it encompasses a broad array of subjects that people with special needs and their families encounter, from estate planning to government benefits to guardianship to advocacy. Attorneys who focus on special needs planning have dedicated their practices to helping families with a wide variety of legal issues, and they must master a vast section of the legal canon in order to properly assist their clients. But all too often, clients arrive at the doorsteps of special needs planners with what they see as specific problems, and they believe that there will be a single, quick solution. Fortunately, good special needs planners don't operate this way, because unlike some types of law that require a concentrated burst of effort to “solve” a particular problem, special needs planning is a marathon, not a sprint.

Of course, there are some times when clients need immediate solutions to very concrete problems, and special needs planners are happy to help. For instance, if someone is seriously injured in an accident and can no longer make decisions for himself, his family may need to pursue a guardianship right away, and this one step may consume significant time and energy, both by the family and by a special needs planner. That's a sprint. But

what happens once a family obtains guardianship? Do they no longer require special needs planning? Of course not!

The guardian will need assistance filing annual reports and accounts with the court, and this could go on for years.

If the injury came about because of someone else's negligence, then the injured party may need a special needs trust to hold a personal injury settlement. If he qualifies, he may need to apply for government disability benefits, including Supplemental Security Income, Social Security Disability Insurance or Medicaid.

Family members may have to change their own estate planning documents to make sure that they reflect their relative's special needs.

No one needs to tell parents of young children with special needs that planning is a marathon (even though daily life may seem like one endless sprint). These families often come to special needs planners because they want to provide for their children if something happens to them, and this often leads to the creation of a special needs trust and a coordinated estate plan for the parents. As the child grows older and reaches the age of majority, he may need a guardianship if he is incapable of managing his own affairs. However, in many cases, the 18-year-old with special needs will be able to sign his own estate planning documents delegating the power to make health care decisions through a health care proxy and

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SPECIAL NEEDS PLANNING (CONTINUED FROM PAGE 9)

naming an attorney-in-fact to assist with financial affairs, and a special needs planner can help him do this. Housing, financial aid for college, and getting ready for independence all take years of planning, and the plans often change over time as an individual grows.

Special needs planners provide all of these services because their focus is on the long-term health and well-being of the client, not just on the immediate issues at hand. That's

why attorneys who focus on this area of law are called "planners."

It can be frustrating to enter an attorney's office with one problem only to realize that there is a lot more to think about (and probably worry about, at least initially) than you thought. But a good special needs planner will set your mind at ease and help you approach the future with confidence, and when emergencies strike (as they often do), your planner can jump to the

rescue with the added benefit of having already gotten to know your family. If you've already started working with a special needs planner, make sure that you stay in touch. And if you're just starting out, welcome to the marathon -- your planner will be there every step of the way.

For additional articles and information from The Academy of Special Needs Planners, go to www.specialneedsanswers.com.

Update on Juvenile Drug Trial

Erika Augustine, MD, Senior Instructor of Neurology, University of Rochester Medical Center

The University of Rochester Batten Center of Excellence, with funding from BDSRA and the Food and Drug Administration, has embarked upon a research project entitled Juvenile Batten Disease Mycophenolate Phase II study (JUMP). This research study will focus on evaluating whether an investigational drug is safe and well tolerated in children with JNCL. Mycophenolate mofetil (also known as Cellcept) is a medication that suppresses the immune system. The study is 22 weeks long with a total of 12 study visits. Four visits require travel to the University of Rochester Medical Center in Rochester, New York. Four visits will be with your child's local physician. Four visits will take place by telephone. Travel costs will be covered by the study. Children enrolled in the study will take mycophenolate syrup twice a day, and will have blood drawn at each study visit.

We have held the first meeting of the board that will monitor safety during the study, have commenced with screening potential participants, and have started the process of setting up site-specific regulatory documents, contracts, and ethics board review for initial sites. We soon anticipate our first formal enrollment. We appreciate the families who have already expressed interest. We will continue to contact these families over time to further review eligibility and to discuss requirements for a local physician to work with us for safety monitoring.

For further information, please contact Amy Vierhile, NP, at (585) 275-4762.

NCL2012

International Conference on Neuronal Ceroid Lipofuscinose

NCL2012 is the 13th International Conference on Neuronal Ceroid Lipofuscinoses (Batten disease) and the 1st Worldwide Meeting for Batten Parents Organizations.

For more information, go to www.ncl2012.org.

March 28 - 31, 2012
Windsor Conference Centre
Royal Holloway
University of London
London



David's Refuge

by Warren and Brenda Pfohl

Two years ago our son David passed away after a thirteen year battle with Batten Disease. Over these two years we have grieved, healed, and prayed for ways to maximize the influence of David's life. As a result we have started a not-for-profit called David's Refuge, Inc.

Our mission is to provide a place of respite for parents and guardians of children with special needs or life threatening medical conditions where they will be refreshed, restored, and re-envisioned in their role as caregivers. Having had the privilege of raising and caring for David we understand the feelings of isolation, exhaustion, and stress families experience in their role as caretakers. We've wrestled with questions of faith, felt the sting of broken dreams, and have questioned whether what we were doing really mattered. What we needed most was just a break where



we could take a deep breath and press on in loving and caring for our family. As our guests you will enjoy the beauty, hospitality and relaxation of a personally designed retreat in a private wing of our home, where

meals, recreation, and opportunities for 'unplugging' are generously provided.

David's Refuge allows parents and other caregivers much needed time to rest, reflect and recharge from the stress of full time care giving. We want every person who stays at David's Refuge to know three things: that you are not alone, what you do matters, and there is a God who loves you. If you want to know more about David's Refuge please check out our website at www.dauidsrefuge.org.

If you have any questions please feel to email us at info@dauidsrefuge.org or call us at (315) 682-4204. We invite anyone who can make it to New York to stay at David's Refuge where you will be reminded that you are not alone, what you do really matters, and that God loves you!



Highlights from BioMarin's Research & Development Day

(Note: Below is an excerpt from BioMarin Pharmaceutical Inc.'s Press Release of December 8, 2012. To read the full press release, go to <http://phx.corporate-ir.net/phoenix.zhtml?c=106657&p=irol-newsArticle&ID=1637904&highlight>.)

NOVATO, Calif., Dec. 8, 2011 / PRNewswire/ -- BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) today hosted a Research and Development Day where members of the company's management team and industry experts provided an update on BioMarin's product portfolio and advancements in the research and development pipeline.

"We believe 2011 has been a year of significant progress in both our late stage and early stage clinical development programs," said Jean-Jacques Bienaime, Chief Executive Officer of BioMarin. "This progress sets the stage for multiple data readouts in the coming year from key programs throughout our pipeline. We believe these events will help move us forward in delivering more therapies that could make large impacts on the lives of patients suffering from several rare diseases."

Program Highlights

BMN-190 for Late infantile neuronal ceroid lipofuscinosis (LINCL) – Form of Batten Disease

At R&D Day, BioMarin also announced a new clinical program, BMN-190 for LINCL, one form of Batten disease. An orphan neurodegenerative disease, LINCL is caused by buildup of lysosomal storage in the CNS. Tripeptidyl Peptidase-1 (TPP1) enzyme deficiency is due to a mutation in the gene CLN2. Neurological symptoms



present between ages two and four, with patients usually confined to wheelchair and blind by around age six. Most patients are deceased between the ages eight and twelve. Incidence is estimated at 3.6 to 4.6 per million births and prevalence is between 350 and 1000 patients worldwide, likely higher due to under diagnosis.

The BMN-190 program is developing a TPP1 enzyme replacement therapy for treatment of LINCL patients. Pharmacological effects, including functional improvement and life extension, have been robustly demonstrated in relevant animal models of LINCL. Pharmacokinetic profile (CSF and plasma) and CNS distribution were favorable after ICV infusion. Toxicity profile is clean after single and repeat ICV infusion administration.

"The BMN-190 program is a great fit in our growing pipeline. LINCL patients represent a significant and unmet medical need in an orphan disease," said Hank Fuchs, M.D., Executive Vice President and Chief Medical Officer of BioMarin. "We believe we can leverage our expertise in biologic manufacturing and proven track record of expeditiously bringing life-altering therapeutics to patients to move this program forward rapidly. We look forward to updating you on advancements in this and other programs in our product pipeline."

About BioMarin

BioMarin develops and commercializes innovative

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HIGHLIGHTS FROM BIOMARIN'S RESEARCH & DEVELOPMENT DAY

(CONTINUED FROM PAGE 12)

biopharmaceuticals for serious diseases and medical conditions. The company's product portfolio comprises four approved products and multiple clinical and pre-clinical product candidates. Approved products include Naglazyme® (galsulfase) for mucopolysaccharidosis VI (MPS VI), a product wholly developed and commercialized by BioMarin; Aldurazyme® (laronidase) for mucopolysaccharidosis I (MPS I), a product which BioMarin developed through a 50/50 joint venture with Genzyme Corporation; Kuvan® (sapropterin dihydrochloride) Tablets, for phenylketonuria (PKU), developed in partnership with Merck Serono, a division of Merck KGaA of Darmstadt, Germany; and Firdapse™ (amifampridine), which has been approved by the European Commission for the treatment of Lambert Eaton Myasthenic Syndrome (LEMS). Product candidates include GALNS (N-acetylgalactosamine 6-sulfatase), which is currently in Phase III clinical development for the treatment of MPS IVA, amifampridine phosphate (3,4-diaminopyridine phosphate), which is currently in Phase III clinical development for the treatment of LEMS in the U.S., PEG-PAL (PEGylated recombinant phenylalanine ammonia lyase), which is currently in Phase II clinical development for the treatment of PKU, BMN 701, a novel fusion protein of insulin-like growth factor 2 and acid alpha glucosidase (IGF2-GAA), which is currently in Phase I/II clinical development for the treatment of Pompe disease, and BMN 673, a poly ADP-ribose polymerase

(PARP) inhibitor, which is currently in Phase I/II clinical development for the treatment of genetically-defined cancers. For additional information, please visit www.BMRN.com. Information on BioMarin's website is not incorporated by reference into this press release.

Forward Looking Statement

This press release contains forward-looking statements about the business prospects of BioMarin Pharmaceutical Inc., including, without limitation, statements about: the expectations of revenue and sales related to Naglazyme; the financial performance of the BioMarin as a whole; the continued clinical development and commercialization of Naglazyme; and actions by regulatory authorities. These forward-looking statements are predictions and involve risks and uncertainties such that actual results may differ materially from these statements. These risks and uncertainties include, among others: our success in the continued commercialization of Naglazyme; our ability to successfully manufacture our products and product candidates; the content and timing of decisions by the U.S. Food and Drug Administration, the European Commission and other regulatory authorities concerning each of the described products and product candidates; the market for Naglazyme; actual sales of Naglazyme; and those factors detailed in BioMarin's filings with the Securities and Exchange

Commission, including, without limitation, the factors contained under the caption "Risk Factors" in BioMarin's 2010 Annual Report on Form 10-K, and the factors contained in BioMarin's reports on Form 10-Q. Stockholders are urged not to place undue reliance on forward-looking statements, which speak only as of the date hereof. BioMarin is under no obligation, and expressly disclaims any obligation to update or alter any forward-looking statement, whether as a result of new information, future events or otherwise.

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The Nurse's Corner:

G-Tubes — When to Insert, Formulas, and Care

by Nancy Carney, RN



This issue's article is about G-tubes, when to insert, formulas, care of G-tubes, etc.

When to make a decision

The decision may be made for you if a modified swallowing study is done and it is apparent that your child is aspirating constantly or develops aspiration pneumonia. It is much better to have a GT/JT (gastrostomy or jejunostomy tubes) inserted when there are no other problems to deal with and do it prophylactically before it really becomes a major issue.

There are two choices – either you interfere and begin some type of supplementary feeding via a tube or you do not interfere and continue as best you can. A supplemental feeding will definitely prolong life and this becomes a major question. There is no right or wrong, and it is important that families openly discuss with a wide variety of professionals the various options that are available. The decision to tube feed is a very difficult one – the realization that there is a loss of skills, perhaps the guilt that you cannot manage some part of this, the prospect of a surgical procedure and pain, and all the other emotionally charged issues, including what it will do to getting people to help care for your child (tube feeding is a “nursing” skill) and what about taking care of my child in school. If you know that you are going to support with supplementary feedings, it is better to begin before there is real nutritional compromise and dehydration. Once you as parents have reached a decision, then it is important to make

sure that the professionals involved in the care of your child are fully aware of what is to be done. If parents decide not to place a feeding tube - which is the decision of some parents – it is because they may feel that by this time in the degenerative process of Batten disease, they no longer want to prolong the life of their child. And also remember that just because you have made the decision to not start a tube feeding by placing a tube and withholding nutrition, you are not withholding other forms of care, especially comfort. But, please, also always remember, that whatever your decision, you will be supported by BDSRA.

Purpose of tube feedings

The primary purposes for the alternative means of tube feedings are: safety, prevention of pneumonia (usually aspiration pneumonia), administration of medication, maintenance of hydration, preserving energy levels, maintaining weight, adequate caloric consumption, and for those children who demonstrate a progressive loss of oral motor skills. The term “feeding tube” has a different meaning for every individual who is faced with the decision of whether or not to pursue this treatment. Again, this is a problem that many families are faced with. It is totally a personal and family decision to insert or not to insert a G-tube. The most important issue here is that you will be supported in your decision. The secret of tube feeding successfully comes in seeing tube feeding in the same light as what would occur if your child were verbally able to tell you about his hunger or thirst at the time you are offering a feeding. There are days when all of us are hungry and days

when our appetite is “off.” Developing an intuitive sense about the total state of well-being is as important as the calories consumed and, even though our ultimate goal may be to add a few pounds, comfort is important.

What kind of formula

You will probably be working with a nutritionist to determine the calories that will be needed for your child. Calculation needs to be made based on height and weight combined with the level of activity – not on age. A normally active nine year-old may require over 2,000 calories a day. Yet a child the same size that is confined to bed may only require 1,000 calories. Some allowance must be made for an increase in tone, or if there is a lot of spasm or perspiration, adjust both calories and fluid amount.

Types of feedings available

Blended food – homemade food which has been blended is seldom used as it is troublesome to prepare and there is a higher chance of causing blockage of the tube and also vomiting of your child, but certainly is an option. If your child is still permitted solid food, mix in a blender, the residue and fiber of a blended normal diet promotes bowel function and most children prefer it to a liquid formula.

Commercial food – can provide complete nutrition for the day, readymade liquid or powder form (cheaper form), additives are available like additional fiber, soy products, etc. There are many different brand names that are available today and you can get them through your health care provider. In the beginning, you may just want to use tube feeding as a supplement to the oral intake. So

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that if certain foods are still enjoyed, these can be fed orally – perhaps your child really enjoys ice cream or pudding, or is a mashed-up spaghetti fan, then let him have what he can handle and make up the liquids and difference with tube feeding. Very often, there has been a weight loss and some slight dehydration before the procedure is done. Advance slowly – let your child set the pace. Clear liquids, beginning with water and then advancing to diluted juices – hydration comes first, then calories. And usually with hydration comes an immediate weight gain, a change in urinary output and sometimes a nice change in the consistency of bowel movements from hard and lumpy to soft, firm consistency. From juices begin at quarter to half to three quarter strength, then you can move to a full-strength formula.

Medications through a feeding tube

In addition to using the tube for feedings, you will be using the tube to give medications. Medications may be administered through a feeding tube utilizing the bolus feeding method. The physician or pharmacist should be asked for liquid medication where possible versus pills or capsules. Formula, juice or milk may be used if the medication does not dissolve in water, but most medications do fine dissolving in water, especially warm water.

Different methods of tube feedings
If your child has an NG in place you will want to check for placement using the following technique. If the feeding tube is secured in place as in a GT/JT, you can do it but it is not a necessity. You also need to be shown how to check for residual

during your teaching of tube feedings and equipment.

Gravity - using a bag – place a stethoscope over the stomach, just to the left of center, kink the feeding tube with your thumb and forefinger to prevent the stomach contents from flowing out, remove the cap and connect a syringe with plunger attached with 5-10cc's of air in it, unkink your tube, quickly insert the air while listening with the stethoscope, you should hear a pop sound which represents air entering the stomach, do not start the feeding if you do not hear the air rush into the stomach.

Continuous feeding – you will want to check placement as above before starting the feeding, fill your bag and tubing with formula before connecting it to the feeding tube, put the tubing with the cassette into the machine (pump), make so it is set at the proper rate and start your feeding.

Bolus feedings – you will want to check for placement and residual as with gravity feedings, you may also insert the plunger and gently push the formula in slowly, make sure you pinch the tubing off before inserting another syringe to avoid excess air getting into the stomach, make sure you rinse the tube with water when the formula is finished. Never force fluids through a tube. Infuse the feeding as slowly as you can to prevent abdominal cramping, nausea, vomiting, gastric distention or diarrhea – if the formula is not infused slowly they are at a higher risk of aspiration and the complications of pneumonia. This method allows more freedom in that you can give feedings anywhere.

Guidelines for giving a tube feeding

Make sure you wash your hands with soap and water before feeding your child. Prior to each feeding, the tube must be checked for patency (to make so line is clear and not blocked) and checking for residual) – the doctor will tell you the level he feels is appropriate for your child – but as a rule, under 150cc's is acceptable, if over that amount, withhold the feeding until the level goes down, you must be certain to reinstall the withdrawn gastric (stomach fluid) contents, to prevent loss of nutrients and electrolytes, and to check the markings (cm) on the tube to be sure it has not moved.

Formula should be given at room temperature (too hot or cold could make your child uncomfortable), unused formula or blenderized foods should be refrigerated and warmed to room temperature before feeding at the next time, but never heat the solution as this could increase the growth of bacteria. Your child should be fed in an upright position (at least 30 degrees) and remain there for 30-60 minutes following the feeding to minimize the possibility of aspiration and its complications.

Care of a feeding tube (Gastrostomy or Jejunostomy Tube [GT/JT])

Each doctor probably has his own specific guidelines for care of the tube. What is listed below is just one possible method of treatment. You will be taught how to care for a gastrostomy/jejunostomy (GT/JT) tube while in the hospital or if done as an outpatient – before you go home.

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Remember this is just one possible method.

- Dressing changes should be done every 1-2 days (every day initially) and include cleansing around the tube with half-strength hydrogen peroxide, a cotton-tipped applicator or a cotton ball works well to remove any crusting or drainage, they need to be clean but not sterile, the hydrogen peroxide may be diluted up to one-half strength with water – just make sure that a fresh swab is used each time rather than placing one that has touched the site back into the solution, be sure to rinse the exit site well and dry the skin completely, apply an antibiotic ointment (not all use this) to the skin around the tube, dress with gauze pads and tape, and position the tube so it does not kink. Remember, if you use a scissors to cut off dressings, be extremely careful not to cut the feeding tube itself. I have seen it cut and then many other problems can occur, worst being surgery and the need to replace the tube. After healing, you can switch to soap and water to cleanse each day.
- Showers are recommended over tub baths to prevent infection at the site, cover the dressing with a double layer of plastic wrap (not all do this) and tape the edges, removing the plastic wrap and change the dressing after the shower.
- Feeding – use water to flush the tube after each feeding, use liquid forms of medications if possible, ask your doctor or nurse to provide

you with specific information about feedings or medications.

- It does not hurt to lie on the tube after the initial healing takes place – in fact, you need to insist that some time be spent on lying prone, if at all physically possible, no restrictions once healed.
- Leakage – the tube may pull away from the abdominal wall resulting in leakage around the insertion site; it may also occur if the incision enlarges in your child with poor nutrition, excessive tension on the tube may cause the tube to be pulled out prematurely. Make sure that the anti-reflux valve is not sticking or is broken – it may need to be replaced. The tube is marked (in cm) where it should be level with the incision and should be checked daily to make sure it has not moved. If it does, call your doctor and he will advise you how to return it to its original position – some parents have said that they had more problems with leakage when a foley was used for the GT/JT in children with Batten disease
- Wound infection – purulent drainage (pus) around the tube is commonly seen but does not always represent a true infection. It may be the body's reaction to a foreign object, such as swelling, tenderness, redness, or drainage of pus around the tube – if an increase in redness occurs, apply an over the counter preparation such as Bacitracin or a cortisone cream. If the redness continues to extend or there is a foul smelling thick drainage (pus), call your doctor. Clean the site more

frequently for a few days, do not use occlusive (air tight) dressings – you want to let some air to the site to promote healing.

- If skin irritation or excoriation (abrasion of the outer skin by trauma, chemicals, burns) is seen at the incision site, apply a skin barrier for protection via a prescription.
- If the tube falls out completely – call your doctor immediately. The tube usually can be easily replaced if it is done within 24 hours from the time it fell out. Waiting longer could mean that a separate new tube may have to be placed.
- Secretions from the stomach irritating the skin – apply an antacid such as Amphojel or Gelusil to prevent redness.
- A lot of movement of the tube, the tube has been pulled out, some bleeding – continue to treat with hydrogen peroxide but increase the number of times per day, use the ointments suggested.
- Pieces of tissue rising above the skin around the tube (granulation) – touch gently with silver nitrate sticks (have the home health nurse or your doctor show you how). This cauterizes the tissue; it will turn black and then peel off and, eventually, the opening should heal flat like a belly button.
- The tube being pulled in by the tugging of the stomach (peristalsis) – pull the tube out to its proper position and secure by wrapping

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the tube with a piece of tape or slitting a baby bottle nipple and taping it onto the tube so that the rounded end will pull against the skin.

- The tube cannot be pulled back to the mark (at the cm mark) and there may be cramps, discomfort, etc. – the tube has probably worked its way into the small intestine. Deflate the balloon (if one) using a syringe (unclamp the tube), pull the tube back to its correct position and then re-inflate the balloon – take a magic marker and make a small mark on the tube or look at the number of centimeters as seen at the skin level when you get home from the hospital, check it daily for the correct position.
- Hissing or flow of liquid around the tube – hissing usually indicates a build-up of gas, unclamp the tube and holding it up to prevent loss of fluid allows the gas to escape, constipation creates pressure also and can force the feeding back

Common tube feeding problems and what to do

This first group includes symptoms that require more immediate attention followed by those that do not:

- Respiratory distress – call for help, initiate emergency plan. Color changes or changes in breathing – may be caused by increased airway secretions, may need suctioning or increase in suctioning. Stop the feeding immediately, check the tube for placement, assess for other problems – possibility of aspiration may have occurred, and follow your child's specific guidelines set for him/her.

- Gagging/Choking /Color changes or changes in breathing when the feeding is not in progress – check for tube placement and assess for other problems.
- If tube falls out – cover stoma (opening) and call family, school nurse, home health nurse and/or doctor, the tube may need reinserted immediately if the tract closes quickly.
- Diarrhea – cause may be too rapid feeding, too concentrated formula, intolerance to formula or medications – if diarrhea occurs, administer small, frequent, less concentrated feedings, make sure the tube feeding is not cold and that proper storage and sanitation procedures have been followed. Skin care may be necessary around perineal (private) area depending on severity of diarrhea slow the feeding/flow rate, dilute the formula with water, gradually increase concentration over 3-5 days, may want to administer Reglan (Metopromide) to increase gastrointestinal (GI) motility if OK with your doctor, warm the formula, for 30 minutes after feeding, position your child on his right side with his head elevated to facilitate gastric emptying. Call your doctor – he may want to reduce the amount of formula being given during each feeding.
- Cramping – formula may be too cold, tube in the wrong place, too fast feeding – use a formula at room temperature.
- Constipation – inadequate fluid provided, low fiber diet, lack of activity – wash down all feedings

with water, provide additional feedings of water if tolerated or prune juice; administer bulk laxatives; fruit, vegetable, or sugar content of feeding may be increased; consult your doctor if constipation continues for more than 3 days.

- Vomiting – too rapid feedings, tube too large, improper tube placement, large residual in stomach (remove residuals as ordered), formula too concentrated, medications given with feeding – slow the feeding; use smaller sized tube; reposition; monitor electrolyte levels if large amounts or continued vomiting, be sure to check your child's specific guidelines, call your doctor – he may want to adjust formula content, to correct deficiency, check for other problems which may contribute to vomiting.
- Nausea – during feeding may indicate delayed gastric emptying, stomach distention, temperature of formula too cold, infusing too fast – stop the feeding, resume feeding when nausea subsides.
- Gastrointestinal reflux – large residual in stomach, physiologic problem – elevate head before, during and 30 minutes after feeding, thickened feedings, and/or giving medications.
- Someone on continuous tube feedings can be in a state of chronic dehydration – make sure they get enough extra fluids.
- Suctioning can cause depletion of electrolytes in the body adding to

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dehydration.

- Aspiration of gastric secretions – discontinue feeding immediately; perform suctioning (nasally or tracheally) of aspirated contents, if possible; notify your doctor – prophylactic antibiotics and chest physiotherapy may be ordered; check tube placement before feeding to prevent complication.
- Tube obstruction – may be due to inadequate flushing of the tube or very thick liquids (formulas) - flush tube with warm water or cranberry juice, if necessary, replace tube; flush tube with 50 ml of water after each feeding to remove excess sticky formula, which could occlude (block) the tube.
- Nasal or pharyngeal irritation or necrosis (injury or death of tissue) – provide frequent oral hygiene, using mouthwash or lemon and glycerin swabs, use Vaseline on cracked lips; change position of tube, if necessary, replace tube.
- Redness, irritation, bleeding, drainage – check to be sure tubing is not being “tugged or pulled,” may be leakage of stomach contents; check to be sure stoma (opening) is clean – could also be a leakage of food issue; food and medications coming in direct contact with skin; refer to child’s specific guidelines; call the family, nurse and doctor.
- Congestive heart failure – (increase of fluid in the lungs or swelling noted in the legs usually first then the arms) monitor your child’s intake and output and respiratory status; reduce flow rate; call your doctor; administer

diuretics as ordered; decrease your child’s fluid intake and enforce bed rest – if your child is having respiratory problems.

- One of the biggest problems why your child may be nauseated or begin to vomit, you might be overfeeding your child. Sometimes, doctors tell you to give your child too much formula. You know your child best. Just because the doctor initially says to give your child 6 cans of formula a day – if he starts vomiting – try reducing the number of feedings or the amount of each feeding and see if the vomiting dissipates.
- Monitor blood and urine glucose (sugar) levels to assess glucose tolerance – monitor serum electrolytes and other blood studies to determine response to therapy – if sugar levels become an issue – notify your doctor to do lab work to check.
- Assess dehydration - may cause constipation – increase fluid intake, may need laxatives or enema.
- Bloating and retention – may be caused by frequent and large volume of feeding at one time.
- Metabolic disturbances – can be caused by dehydration, diarrhea, or vomiting.
- Glycosuria, cramping, abdominal distention indicate intolerance of feedings.
- Dumping Syndrome - definition of dumping syndrome is when the stomach’s contents empty too rapidly into the intestines causing symptoms. If the tube is placed

directly into the jejunum (J-Tube), the chance of dumping syndrome is increased due to the food already entering the body directly into the small intestine, and then the food goes too rapidly through the intestine where most of the absorption of nutrients takes place

Documentation

- Tube feeding sheet – to be completed after each feeding and indicate the date, time feeding began, rate, type and amount of feeding, and the flush, skin care, tube placement check and residual check.
 - Intake/output record – as indicated, weight record – weigh as ordered. Vital signs sheet – as indicated.
 - Bowel movement chart/daily monitoring record.
 - Communication book – as indicated.
 - Notes – include events that occurred such as any time a continuous pump feeding is shut off and the time that it is turned back on. Observation for placement and color/amount of volume, distention and/or discomfort during feeding, vomiting or diarrhea, any other unusual symptom for this particular child.

There is much more information in the Gastrointestinal Book that is available from BDSRA. If you have any questions, please contact the office.

Kid's Corner

by Donna Gunn, Office Manager

BDSRA would like to give a **BIG “Hip, Hip Hurray!”** to the young people of families affected by Batten disease who are willing to give of their time and generosity of heart to help in the fight against this disease. We graciously thank:

William Nogle, after hearing that Batten disease had taken his cousin, Jemyni Bean, initially wanted to sell all of his toys to donate to the Batten disease cause. Instead, 8 year-old William and his sister, 6 year-old Meegan, held a lemonade stand fundraiser to raise money in memory of Jemyni. What a wonderful way to show your love for a cousin and to continue the battle against this disease for other children!

The Freshman Class of Edison High School in Berlin Heights, Ohio, decided to collect family recipes from relatives and friends to create a cookbook entitled “Every Moment Counts” to raise funds for Batten disease in honor of their classmate, Asher Nikolajevs. Class of 2015...you all are AWESOME!!

Since 10 year-old **Liam Jorgenson** of Oro Valley, Arizona, could not actually be at the Barkin' For Batten event in Columbus, Ohio, on October 15, Liam decided to participate by borrowing Snoopy, a neighbor's dog he's known since puppyhood, to walk and raise money for BDSRA. Big brother, Noah, helped Liam go door-to-door for sponsors and helped him prepare and distribute thank-you cards. Great job, Liam and Noah!

Zia Jenkins, a senior at Cape Cod Regional Technical High School in Harwich, Mass., did her senior project on Batten disease and, as part of the project, she raised money to donate to BDSRA. She, too, has been personally touched by this disease. Her older brother, Barry, passed away from Batten disease and Zia is determined to get the word out. We need so many more like you, Zia!

Caroline and Anna Medley, and their friend, Grace Schinsing, put their thoughts together and came up with a new version of the lemonade business by putting together the “Fresh Squeezed Juice Sale,” collecting money in honor of their brother and friend, Jake Medley. What a super idea - you are definitely special young ladies!

In honor of Miss Celia Betz, the staff and students of **Holt Crossing Intermediate School** in Grove City, Ohio, sold wristbands and donated money to help fund research. The school staff and all of us here at

BDSRA are very proud of these students' generosity and their spirit of getting involved to help others.

The 6th Grade Class of Riverside Presbyterian Day School in Jacksonville, Florida, wanted to do a fundraiser and Clara Trednick, a longtime friend of the Medley Family, suggested that BDSRA would be a good charity to support (thank you, Clara!). The students gathered all the books they could find and held a used book sale...what a “novel” idea! Not only have they given to the Batten disease fight, but they helped to spread the knowledge of those books to others.

It is amazing to see the compassion, sincerity and drive of the young people of today and serves as great encouragement - not only for the eradication of Batten disease, but for our world as a whole.

We are so proud of all these young people and THANK them from the depths of our hearts!



Diseases Can Affect More Than Just The Infected

By Caitlin Bishop (Note: Caitlin wrote this paper about her aunt, Linda Sivulka, who had JNCL, for a college course in September 2011.)

When you think of neurodegenerative diseases, what do you think of? Do you even know what the word actually means? Maybe you think of Parkinson's or Alzheimer's because those are a few of the most popular. Those diseases never really mattered to my family; the one that does matter to us is Batten's Disease. Just to give a bit of a mental picture of what this disease is, on bdsra.org, the official Batten's Disease website, it states that "Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills." But this isn't all that Batten's Disease encompasses.

Although only one person in my family, my aunt, had the disease, it has deeply affected all of us in some way. The effect that it had on my aunt was obviously much different than the effect that it had on me or my mom, but experiencing the disease does more than just harm the person who is living with it. To me, Batten's Disease is what took my aunt. And it could take others in my family as well, and we would never know until it was too late. There is no cure, and researchers have not yet figured out which gene is the infected one. It's a scary thought, but even I could be a carrier for the disease and I could pass it on to my children. To my aunt, the disease is what kept her from being able to do many of the things she loved the most and sometimes kept her from understanding why she couldn't do those things anymore.

My experience with this disease has taught me many things. I feel like I am put into a category that most people my age are not. I can work with people with disabilities and not want to make

fun of them or be afraid of them. A physical or mental disease does not define a person, and sometimes people who haven't been in a situation like mine can't realize that. It's the same for me. I'm not defined by the disease that took my aunt, but it still played a huge part in my life and will continue to do so for the rest of my life. It played a huge part in the lives of all my family members. We all were, and still are, affected in different ways from our experience with this, so I can only speak of my personal experience. I will in no way, shape, or form say that Batten's Disease affected my life in a positive way, because I would never wish it to happen to me or anyone else ever again, but looking at the positive side of the situation is how I can accept and learn from my experience. Batten's Disease has taught me tolerance and understanding, compassion and patience.

My aunt, on the other hand, obviously had a different experience than I did. Of course, I cannot know exactly how her experience was, but from spending time with her and seeing her day to day, I have some sort of idea how it affected her. Batten's Disease took so many things away from my aunt, things that she once loved to do. She loved to swim, but as the disease progressed, her growing immobility caused her to not be able to swim as much. It was the only time she could be out of her wheel chair and not need constant help to do everything. Instead of walking, she could swim. A few family members took her to swim a couple of times but slowly, even swimming became impossible. It wasn't that she was completely immobile, but that she couldn't walk. She could still use and move the

upper half of her body, but most of even that movement was shaky and not stable. Along with swimming, my aunt also had a big love for shopping. It didn't matter what kind of shopping, she loved any kind. My grandma used to take her with to her job at Target during the day and my grandpa would pick her up on his way home. While my grandma was working, she could spend hours on end just roaming stores, sometimes not even buying anything. But again, as the disease progressed, it was harder and harder to take her to stores without something happening. Since her upper body movements were shaky, she couldn't look at many things without dropping or hitting other items. Aside from swimming and shopping, my aunt loved card games and seeing all of her little nieces, nephews and all other children in our family. She had the biggest heart of anyone I've ever known, and that was the one thing that the disease could never take from her. She never stopped loving everyone and everything.

Batten's Disease doesn't affect as many people as Parkinson's Disease or Alzheimer's, but that doesn't mean that it's uncommon. The people that are aware of the disease know that it affects many, and the odds are against the people that have it. My aunt passed away on May 9, 2007 when I was in eighth grade and I will always remember it as if it were yesterday. It's been over four years now that she's been gone and I still think about her and think of all the wonderful things she taught me and all of the joy she brought into my life.

♣ In Loving Memory ♡

HAYLEE JOYCE, daughter of Chris & Clarissa Joyce, Blue Springs, MO
Born: 03-02-07 ♦ Died: 12-10-11 ♦ Infantile

BOONE BARRICKLOW, son of Douglas & Cathy Barricklow, Jasper, MI
Born: 12-20-86 ♦ Died: 12-07-11 ♦ Juvenile

BLAKE JAEGER, son of Jeremy & Dawn Jaeger, Granite Bay, CA
Born: 01-11-02 ♦ Died: 11-21-11 ♦ Late Infantile

MATHEUS A. de SOUZA dos ANJOS, son of Joao Roberto dos Anjos, Paraná, Brazil
Born: 05-02-99 ♦ Died: 11-19-11 ♦ Late Infantile

DONOVAN LUSK, son of Darrin & Amanda Lusk, Italy, TX
Born: 11-27-02 ♦ Died: 11-11-11 ♦ Late Infantile

WILLIAM PHELAN, son of Jeff & Diane Phelan, Las Vegas, NV
Born: 02-20-02 ♦ Died: 11-09-11 ♦ Late Infantile

MARIAH MALLON, daughter of Bonnie Mallon, Fond du Lac, WI
Born: 11-04-94 ♦ Died: 10-11-11 ♦ Juvenile

DANIELLE BUSBY, daughter of Mike & sister of Brittany Robinson (JNCL), Surprise, AZ
Born: 03-31-74 ♦ Died: 10-11-11 ♦ Unaffected Sibling

MICHAEL PANKRATZ, son of Tim & Martha Pankratz, Tucson, AZ
Born: 02-14-86 ♦ Died: 09-27-11 ♦ Unknown Confirmed NCL

SEAN NEWELL, son of Bob & Marge Newell, Maple Valley, WA
Born: 03-07-90 ♦ Died: 09-25-11 ♦ Juvenile

ALEXANDER BRAND, son of Robin Brand, Innisfail, Alberta, Canada
Born: 03-06-03 ♦ Died 09-20-11 ♦ Infantile



MATTHEW ALWYN de VILLIERS, son Alwyn & Elva de Villiers, Pretoria, South Africa
Born: 08-24-04 ♦ Died: 09-12-11 ♦ Infantile

CRAIG JONES, son of Ray & Leona Jones, Victoria, Australia
Born: 01-26-75 ♦ Died: 09-03-11 ♦ Kufs

ALEKSANDRA KRASNIAK, daughter of Grey & Bozena Krasniak, Elk Grove Village, IL
Born: 02-05-02 ♦ Died: 09-03-11 ♦ Late Infantile

STEPHEN B. HATCHETT, son of Randy & Sherry Hatchett, Madison, TN
Born: 11-25-85 ♦ Died: 08-23-11 ♦ Juvenile

STEVIE McCUTCHEON, daughter of Glen & Erin McCutcheon,
Dannevirke, New Zealand
Born: 02-08-07 ♦ Died: 07-31-11 ♦ Late Infantile

GEORGE DOUMITT, son of Charles & Margaret Doumitt, Jackson, TN
Born: 03-14-76 ♦ Died: 07-17-11 ♦ Juvenile

BRYCE DALEY, son of Jonathon & Lorraine Daley, Paralowie, S. Australia
Born: 05-13-01 ♦ Died: 05-29-11 ♦ Late Infantile

ARENE SANGRONIZ REMIRO, daughter of Nestor Sangroniz & Arantza Remiro,
Madrid, Spain
Died: 05-28-11 ♦ Late Infantile

JOEY MILANI, son of Bill & Cindy Milani, Frederick, MD
Born: 01-18-93 ♦ Died: 04-15-11 ♦ CLN7

HEATHER SELPH, daughter of Ruth Giacalone, Salisbury, MD
Born: 12-10-84 ♦ Died: 04-05-11 ♦ Juvenile

CLAY CUNNINGHAM, son of Dave & Jane Cunningham, Birmingham, AL
Born: 10-08-02 ♦ Died: 04-05-11 ♦ Juvenile

FOURTH QUARTER DONOR GIFTS

(GIFTS GIVEN SEPTEMBER 13 THROUGH DECEMBER 9, 2011)

Batten Disease Support and Research Association has been remembered many times in the past three months by families and friends of children with Batten disease. To all of you, we express our deepest appreciation for your generous gifts. We sincerely apologize if there are any omissions or misspellings; please alert us to any changes. We kindly ask that with any future gifts, you specifically indicate whether the donation is in "Honor of" or in "Memory of."

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Dad

DANIELLE BUSY – Devoted Sister of Brittany Robinson

Dad

Mr. Peter Black

Mr. Richard Ernst

Mr. Larry Garfinkel

Mr. Mark Heald

Continued on **page 23**

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CONFERENCE:

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SPECIAL EVENTS:

Alabama BDSRA Chapter Golf Tournament
Asher Bash — Honor of Asher Nikolajevs
Freshman Class, Edison High School (Berlin Heights, OH)
Barkin' for Batten
Bash for Batten — Memory of Ben Wempner
Battin' for Batten — Honor of Nathan Hoover
Ms. Sue Hoover
Big House Big Heart 5K
Boogie for Batten Disease Zumba Party
Boston's Battle
Celia's Walk
Come Together for Kaitlin
Cribbage Board Sales — Honor of Kelsey Shuros
Fresh Squeezed Juice Sale — Honor of Jake Medley
Give Back Night — Memory of Dominic Ceravone for his 16th Birthday
Hayden's Hope 5K Run/Walk

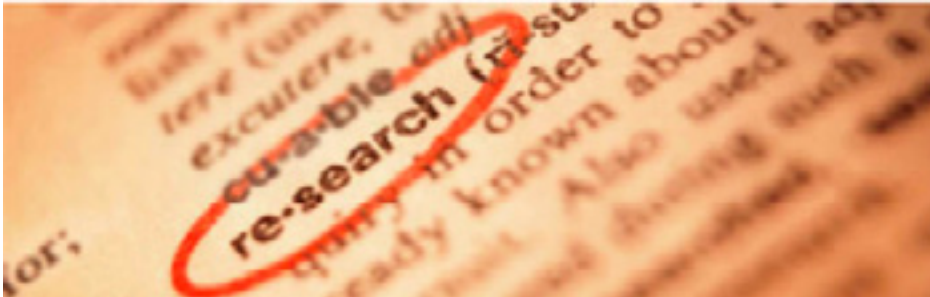
Kennedy Poker Run

Red Land Alumni Association
Kufs Bicycle Run
Lemonade & Cupcake Stand — Memory of Jemyni Bean
Metro New York/New Jersey 2011 Bowl-A-Thon
Mike's Beer Bucket Annual Golf Tournament
PACS Industries
Miles for Michael
The Morgante Fundraiser — Memory of Billy & Joey Milani
Party in the Park — Honor of Jeff Montavon
Riverside Presbyterian Day School, 6th Grade (Jacksonville, FL)
Run for Research (Noah's Run)
Team Rachel's Road Race
Vic's Mud Bog Wine Tasting for Batten Disease — Honor of Jake Medley



Marking NCL Research Progress Through Scientific Reporting

By Danielle M Kerkovich, PhD, Principal Scientist, Beyond Batten Disease Foundation and Scientific Officer, BDSRA



It has been a banner year in Neuronal Ceroid Lipofuscinosis (NCL) research. Clinician scientists and investigators have traveled the globe presenting their findings at scientific meetings and invitation-only conferences, and have published over 70 scientific peer-reviewed articles reporting new research in the 9 forms of NCL and its 10 causal genes.

Below is a brief overview of some of the scientific clues discovered in 2011. Interestingly but not surprisingly, a large number of studies completed this year resulted in findings applicable to more than one form of NCL. Similarity in symptoms and disease progression between the various forms suggest that these diseases share common cellular (dys)functional pathways. If this is true, then therapies discovered in one form of NCL or even other forms of neurodegenerative disease could be used to treat or learn more about another, accelerating our search for a cure in all forms of NCL.

Clues in the Neuronal Ceroid Lipofuscinosis Puzzle: The Search for Drug Targets

A great deal of progress has been made in the arduous search for potential drug targets in NCL, a

key step in the path to finding a treatment for the disease. In February, investigators at Peking University Center of Medical Genetics in Beijing, China, published their analysis of skin cells (fibroblasts) from children with Infantile, Late Infantile and Juvenile NCL and found disturbances in several processes that are important for cell structure and transport of cellular components. Key proteins involved in these processes were also found in different quantities among the various NCLs, being normal in some forms while too much or too little was found in others. It will be important to follow up on these findings to tease apart which cellular disturbances are primary or secondary to disease and whether any of these key proteins can be used as biomarkers of disease progression and someday; biomarkers of the effects of treatment.

Also in February, researchers at Harvard University published the examination of cell lines derived from animal models of variant late-infantile NCL caused by CLN6 mutations and Juvenile NCL caused by CLN3 mutations to learn that while mutations of the CLN6 and CLN3 genes trigger different processes, these processes act like highway entrance ramps to the

same pathway one that is responsible for proper subunit C protein turnover (the major component of accumulated storage protein) and neuronal cell survival.

In September, investigators at the Cleveland Clinic described their findings of a 4-year old boy with Late Infantile NCL. When treated with Levodopa, which is converted to the brain chemical dopamine in the body and is used to treat Parkinson's disease, the child's walking ability improved. Taken together with a previous study showing similar results in a child with Juvenile NCL and work in glutamate receptor activity published by investigators at the Sanford Children's Health Research Center in South Dakota, these results suggest supplementation or modulation of brain chemical activity could be a means of treating secondary symptoms in Infantile, Late Infantile, and Juvenile NCL.

In June, a second *Science* paper from the Telethon Institute of Genetics and Medicine (TIGEM), Federico II University, Naples, Italy and Texas Children's Hospital in Houston, Texas, began to look at pharmacological approaches to modulating the amount and location of transcription factor EB (TFEB). This group previously published their discovery that TFEB increases the number of lysosomes (recycling centers) in a cell and improves the cell's overall ability to degrade accumulated storage material. This second article demonstrates that TFEB, which normally sits outside of the nucleus, can be triggered by

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protein kinase inhibitors to move into the nucleus, turn on multiple genes, and make extra lysosomes. Why is this important? Children with Infantile, Late Infantile and Juvenile NCL have lysosomes that work. They just work poorly and are eventually overwhelmed, resulting in cell death. Therefore, if investigators can make more TFEB or up the activity of existing TFEB so that they make extra lysosomes, then a cell struggling with 5 lysosomes working at 10% capacity could become healthier if it had, for example, 20 lysosomes working at 10% capacity. This would be true not just for neurons, but for other cells in the brain that might suffer from a lack of normal NCL proteins, or heart cells that researchers believe may become damaged in NCLs over the lifespan. The beauty of TFEB is that it could prompt the cell to fix some of its own problems.

October was a busy publishing time for NCL investigators, as 17 of the 72 publications for the year came out during this month. Investigators at the Washington University School of Medicine in St Louis, Missouri, together with collaborators in Sweden, published their findings showing that disrupting structural proteins in the brains of Infantile NCL mouse models accelerates the disease, suggesting that cytoskeletal proteins inside glial cells (cells in the brain that neighbor and support neurons) may be neuroprotective. This is an important finding because it uncovers the brain's natural ability to fend off disease progression, potentially opening up opportunities to treat disease by helping boost the brain's emergency response system.

When investigators look at multiple drug targets inside a neuron, they

approach the problem from several angles, increasing the overall possibility of hitting the right target that will cure the disease. At the same time, researchers are beginning to engage in “drug discovery,” an endeavor previously undertaken only by pharmaceutical companies like Johnson & Johnson or Pfizer. Luckily, that has changed. Advancements in drug screening technologies are making it possible for scientists to test thousands of existing drugs on disease-affected cells to see if any current compounds have a therapeutic effect on a disease other than the one for which they were originally made. This tactic appears to pay off: 40% of the 49 “new” medications in 2010 were actually existing drugs repurposed for new uses. A few of the drugs reported to have a positive effect on animal models of NCL or human cells are described here.

Teaching Old Drugs New Tricks

Investigators at the National Institutes of Health in Washington, DC, and Hoseo University in the Republic of Korea explored the efficacy of Resveratrol (RSV), an antioxidant found in the skin of red grapes that is under investigation in Alzheimer's disease, mild brain injury, and Multiple Sclerosis. Investigators have reported RSV reduced the damaging effects of oxidative stress in the cells of children with Infantile and Juvenile NCL.

Mycophenolate mofetil (MMF, CellCept) contains a drug that the body metabolizes into a compound that depletes guanosine nucleotides, the ingredients needed to make immune cells. MMF also inhibits the proliferation of these cells, their recruitment to sites of inflammation, the recruitment of other immune cells (monocytes,) and the production of

nitric oxide, which has been shown to have tissue-damaging effects. MMF is FDA-approved to prevent the rejection of transplanted organs. It was originally used to treat psoriasis in the 1970s, and recently it has been tested with other inflammatory conditions like lupus. Researchers at the University of Rochester School of Medicine and Dentistry in Rochester, New York, and King's College London demonstrated mice without CLN3 protein treated with MMF develop symptoms of JNCL-like disease much later than untreated mice. These results suggest that MMF inhibits disease progression by inhibiting the body's immune response (which, paradoxically, may actually make Juvenile NCL worse) or it affects the disease via an as-yet unknown mechanism. The much-anticipated safety and tolerability study of short-term administration of MMF in children with JNCL began at the University of Rochester School of Medicine and Dentistry in July.

Using a cell culture model of Juvenile NCL, investigators at Harvard University showed that Lithium, which has therapeutic effects in a variety of neuronal disease animal models for stroke and Huntington's disease and is currently used for bipolar disease in humans, rescues the impaired cellular recycling program in a Juvenile NCL mouse model and reduces disease-specific neuronal cell death.

Conclusion

As you can see, many NCL researchers around the globe are working together to identify cellular drug targets for NCL while others are exploring the possibility that existing drugs may strike those and unknown drug targets in NCL. This is one of those times that we think burning the candle from both ends is a good thing!

NCL Publications in 2011

January

[pH-dependent localization of Btn1p in the yeast model for Batten disease.](#)

Wolfe DM, Padilla-Lopez S, Vitiello SP, Pearce DA.
Dis Model Mech. 2011 Jan;4(1):120-5. Epub 2010 Oct 19.

[Immunosuppression alters disease severity in juvenile Batten disease mice.](#)

Seehafer SS, Ramirez-Montealegre D, Wong AM, Chan CH, Castaneda J, Horak M, Ahmadi SM, Lim MJ, Cooper JD, Pearce DA.
J Neuroimmunol. 2011 Jan;230(1-2):169-72.

[A novel interaction of CLN3 with nonmuscle myosin-IIb and defects in cell motility of Cln3\(-/-\) cells.](#)

Getty AL, Benedict JW, Pearce DA.
Exp Cell Res. 2011 Jan 1;317(1):51-69. Epub 2010 Sep 17.

[A missense mutation in canine CLN6 in an Australian shepherd with neuronal ceroid lipofuscinosis.](#)

Katz ML, Farias FH, Sanders DN, Zeng R, Khan S, Johnson GS, O'Brien DP.
J Biomed Biotechnol. 2011;2011:198042. Epub 2010 Dec 22.

[Different early ER-stress responses in the CLN8\(mnd\) mouse model of neuronal ceroid lipofuscinosis.](#)

Galizzi G, Russo D, Deidda I, Cascio C, Passantino R, Guarneri R, Bigini P, Mennini T, Drago G, Guarneri P.
Neurosci Lett. 2011 Jan 25;488(3):258-62. Epub 2010 Nov 19.

February

[A knock-in reporter mouse model for Batten disease reveals predominant expression of Cln3 in visual, limbic and subcortical motor structures.](#)

Ding SL, Tecedor L, Stein CS, Davidson BL.
Neurobiol Dis. 2011 Feb;41(2):237-48. Epub 2010 Sep 25.

[Juvenile neuronal ceroid-lipofuscinosis: clinical and molecular investigation in a large family in Brazil.](#)

Valadares ER, Pizarro MX, Oliveira LR, Amorim RH, Pinheiro TM, Grieben U, Santos HH, Queiroz RR, Lopes Gde C, Godard AL.
Arq Neuropsiquiatr. 2011 Feb;69(1):13-8.

[Temporary inhibition of AMPA receptors induces a prolonged improvement of motor performance in a mouse model of juvenile Batten disease.](#)

Kovács AD, Saje A, Wong A, Szénási G, Kiricsi P, Szabó E, Cooper JD, Pearce DA.
Neuropharmacology. 2011 Feb-Mar;60(2-3):405-9. Epub 2010 Oct 29.

[Screening for calcium channel modulators in CLN3 siRNA knock down SH-SY5Y neuroblastoma cells reveals a significant decrease of intracellular calcium levels by selected L-type calcium channel blockers.](#)

An Haack K, Narayan SB, Li H, Warnock A, Tan L, Bennett MJ.
Biochim Biophys Acta. 2011 Feb;1810(2):186-91. Epub 2010 Oct 7.

[A two-dimensional protein fragmentation-proteomic study of neuronal ceroid lipofuscinoses: identification and characterization of differentially expressed proteins.](#)

Wang P, Ju W, Wu D, Wang L, Yan M, Zou J, He B, Jenkins EC, Brown WT, Zhong N.
J Chromatogr B Analyt Technol Biomed Life Sci. 2011 Feb 15;879(5-6):304-16. Epub 2010 Dec 23.

Continued on **page 28**

February (continued)

Distinct early molecular responses to mutations causing vLINCL and JNCL presage ATP synthase subunit C accumulation in cerebellar cells.

Cao Y, Staropoli JF, Biswas S, Espinola JA, Macdonald ME, Lee JM, Cotman SL.
PLoS One. 2011 Feb 17;6(2):e17118.

Disruption of adaptive energy metabolism and elevated ribosomal p-S6K1 levels contribute to INCL pathogenesis: partial rescue by resveratrol.

Wei H, Zhang Z, Saha A, Peng S, Chandra G, Quezado Z, Mukherjee AB.
Hum Mol Genet. 2011 Mar 15;20(6):1111-21. Epub 2010 Dec 28.

[Detection of vacuolated peripheral blood lymphocytes in screening for and diagnosis of lysosomal storage diseases].

Chang XZ, Liu JY, Wu Y, Jiang YW, Xiong H, Wang S, Qin J.
Zhonghua Er Ke Za Zhi. 2011 Feb;49(2):135-8. Chinese.

Lithium rescues the impaired autophagy process in CbCln3(Δ ex7/8/ Δ ex7/8) cerebellar cells and reduces neuronal vulnerability to cell death via IMPase inhibition.

Chang JW, Choi H, Cotman SL, Jung YK.
J Neurochem. 2011 Feb;116(4):659-68. doi: 10.1111/j.1471-4159.2010.07158.x. Epub 2011 Jan 19.

A novel CLN2/TPP1 mutation in a Chinese patient with late infantile neuronal ceroid lipofuscinosis.

Wang YL, Zeng ZY, Song XW, Hao ZF, Shi YW, Tang B, Chen SQ, Gao MM, Di W, Long YS, Yi YH, Liao WP.
Neurogenetics. 2011 Feb;12(1):93-5. Epub 2010 Sep 7. No abstract available.

Interactions of the proteins of neuronal ceroid lipofuscinosis: clues to function.

Getty AL, Pearce DA.
Cell Mol Life Sci. 2011 Feb;68(3):453-74. Epub 2010 Aug 1. Review.

March

Case records of the Massachusetts General Hospital. Case 8-2011. A 32-year-old woman with seizures and cognitive decline.

Sims KB, Cole AJ, Sherman JC, Caruso PA, Snuderl M.
N Engl J Med. 2011 Mar 17;364(11):1062-74. No abstract available.

The specific loss of GnRH-positive neurons from the hypothalamus of sheep with CLN6 neuronal ceroid lipofuscinosis occurs without glial activation and has only minor effects on reproduction.

Kay GW, Jay NP, Palmer DN.
Neurobiol Dis. 2011 Mar;41(3):614-23. Epub 2010 Nov 24.

April

Comment on "Deletion of btn1, an orthologue of CLN3, increases glycolysis and perturbs amino acid metabolism in the fission yeast model of Batten disease".

Pearce DA, Padilla-Lopez S.
Mol Biosyst. 2011 Apr;7(4):1347-8; author reply 1349. Epub 2011 Jan 24.

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Salek RM, Pears MR, Cooper JD, Mitchison HM, Pearce DA, Mortishire-Smith RJ, Griffin JL.
J Biomol NMR. 2011 Apr;49(3-4):175-84. Epub 2011 Apr 3.

[Cardiac involvement in juvenile neuronal ceroid lipofuscinosis \(Batten disease\).](#)

Ostergaard JR, Rasmussen TB, Mølgaard H.
Neurology. 2011 Apr 5;76(14):1245-51.

[BEHAVIORAL DIFFERENCES IN A MOUSE MODEL OF BATTEN](#)

www.tcnj.edu/~joss/2011/2011%20Wall.pdf

Wall, CM

The College of New Jersey Journal of Student Scholarship (TCNJ) Vol XIII April 2011

May

[Btn3 is a negative regulator of Btn2-mediated endosomal protein trafficking and prion curing in yeast.](#)

Kanneganti V, Kama R, Gerst JE.
Mol Biol Cell. 2011 May 15;22(10):1648-63. Epub 2011 Mar 25.

[The Batten disease gene CLN3 is required for the response to oxidative stress.](#)

Tuxworth RI, Chen H, Vivancos V, Carvajal N, Huang X, Tear G.
Hum Mol Genet. 2011 May 15;20(10):2037-47. Epub 2011 Mar 3.

[Reply to Comment on "Deletion of btn1, an orthologue of CLN3 ...](#)

pubs.rsc.org/en/content/articlelanding/2011/mb/c0mb00300jCached

Mole, SE and Codlin, S and Griffin, JL

MOL BIOSYST , 7 May 2011 (4) 1349 - 1349.

[Neuronal ceroid lipofuscinosis in Qatar: report of a novel mutation in ceroid-lipofuscinosis, neuronal 5 in the Arab population.](#)

Al-Kowari MK, Hassan S, El-Said MF, Ben-Omran T, Hedin L, Mole SE, Badii R.
J Child Neurol. 2011 May;26(5):625-9. Epub 2011 Mar 29.

[Kufs disease, the major adult form of neuronal ceroid lipofuscinosis, caused by mutations in CLN6.](#)

Arsov T, Smith KR, Damiano J, Franceschetti S, Canafoglia L, Bromhead CJ, Andermann E, Vears DF, Cossette P, Rajagopalan S, McDougall A, Sofia V, Farrell M, Aguglia U, Zini A, Meletti S, Morbin M, Mullen S, Andermann F, Mole SE, Bahlo M, Berkovic SF.
Am J Hum Genet. 2011 May 13;88(5):566-73. Epub 2011 May 5.

[Altered sensitivity of cerebellar granule cells to glutamate receptor overactivation in the Cln3\(\$\Delta\$ ex7/8\)-knock-in mouse model of juvenile neuronal ceroid lipofuscinosis.](#)

Finn R, Kovács AD, Pearce DA.

Neurochem Int. 2011 May;58(6):648-55. Epub 2011 Feb 17.

Continued on **page 30**

NCL PUBLICATIONS IN 2011 *(CONTINUED FROM PAGE 29)*

June

[Therapeutic approaches to the challenge of neuronal ceroid lipofuscinoses.](#)

Kohan R, Cismondi IA, Oller-Ramirez AM, Guelbert N, Anzolini TV, Alonso G, Mole SE, de Kremer DR, de Halac NI. *Curr Pharm Biotechnol.* 2011 Jun;12(6):867-83. doi: 1389-2010/11 \$58.00+.00. Review.

[TFEB links autophagy to lysosomal biogenesis.](#)

Settembre C, Di Malta C, Polito VA, Garcia Arencibia M, Vetrini F, Erdin S, Erdin SU, Huynh T, Medina D, Colella P, Sardiello M, Rubinsztein DC, Ballabio A. *Science.* 2011 Jun 17;332(6036):1429-33. Epub 2011 May 26.

[A truncating mutation in ATP13A2 is responsible for adult-onset neuronal ceroid lipofuscinosis in Tibetan terriers.](#)

Farias FH, Zeng R, Johnson GS, Wininger FA, Taylor JF, Schnabel RD, McKay SD, Sanders DN, Lohi H, Seppälä EH, Wade CM, Lindblad-Toh K, O'Brien DP, Katz ML. *Neurobiol Dis.* 2011 Jun;42(3):468-74. Epub 2011 Feb 26.

[Challenging symptom profiles of life-limiting conditions in children: a survey of care professionals and families.](#)

Malcolm C, Forbat L, Anderson G, Gibson F, Hain R. *Palliat Med.* 2011 Jun;25(4):357-64. Epub 2011 Jan 12.

[Inborn errors of metabolism for child neurology residents.](#)

Patterson MC. *Semin Pediatr Neurol.* 2011 Jun;18(2):95-7.

August

[Clarifying lysosomal storage diseases.](#)

Schultz ML, Tecedor L, Chang M, Davidson BL. *Trends Neurosci.* 2011 Aug;34(8):401-10. Epub 2011 Jun 30.

[Lentiviral-mediated gene transfer to the sheep brain: implications for gene therapy in Batten disease.](#)

Linterman KS, Palmer DN, Kay GW, Barry LA, Mitchell NL, McFarlane RG, Black MA, Sands MS, Hughes SM. *Hum Gene Ther.* 2011 Aug;22(8):1011-20. Epub 2011 May 19.

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